macrozoospermia

Macrozoospermia is a condition that affects only males. It is characterized by abnormal sperm and leads to an inability to father biological children (infertility).

In affected males, almost all sperm cells have abnormally large and misshapen heads. The head of the sperm cell contains the male's genetic information that is to be passed on to the next generation. Normally, the head of a sperm cell contains one copy of each chromosome. In men with macrozoospermia, the sperm cell head contains extra chromosomes, usually four copies of each instead of the usual one. This additional genetic material accounts for the larger head size of the sperm cell. Additionally, instead of having one tail (flagellum) per sperm cell, affected sperm have multiple flagella, most often four.

Because of the additional genetic material, if one of these abnormal sperm cells combines with an egg cell, the embryo will not develop or the pregnancy will result in miscarriage.

Frequency

Macrozoospermia is estimated to affect 1 in 10,000 males in North Africa. The prevalence of the condition outside this region is unknown.

Genetic Changes

Mutations in the *AURKC* gene cause macrozoospermia. The *AURKC* gene provides instructions for making a protein called aurora kinase C. This protein is abundant in male testes, which are the male reproductive organs in which sperm are produced and stored. In the testes, this protein regulates the division of sperm cells. Aurora kinase C ensures that the mechanisms for cell division are in place and helps chromosomes properly align with each other so that every new sperm cell contains one copy of each chromosome after cell division.

AURKC gene mutations that cause macrozoospermia lead to the production of a nonfunctional protein or a protein that is quickly broken down. This lack of aurora kinase C blocks cell division in sperm cells. Without cell division, the chromosomes are not split among multiple new sperm cells. As a result, affected sperm cells contain extra chromosomes, usually four copies of each instead of the usual one.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal

recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- infertility associated with multi-tailed spermatozoa and excessive DNA
- large-headed multiflagellar polyploid spermatozoa
- spermatogenic failure 5

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Infertility associated with multi-tailed spermatozoa and excessive DNA
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0403812/

Other Diagnosis and Management Resources

- American Society for Reproductive Medicine: Sperm Analysis
  http://www.reproductivefacts.org/topics/detail.aspx?id=1713
- Centers for Disease Control and Prevention: Infertility FAQs
  https://www.cdc.gov/reproductivehealth/Infertility/
- MedlinePlus Encyclopedia: Semen Analysis
  https://medlineplus.gov/ency/article/003627.htm
- MedlinePlus Health Topic: Assisted Reproductive Technology
  https://medlineplus.gov/assistedreproductivetechnology.html

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Semen Analysis
  https://medlineplus.gov/ency/article/003627.htm
- Health Topic: Assisted Reproductive Technology
  https://medlineplus.gov/assistedreproductivetechnology.html
- Health Topic: Male Infertility
  https://medlineplus.gov/maleinfertility.html

Genetic and Rare Diseases Information Center

- Macrozoospermia
  https://rarediseases.info.nih.gov/diseases/12385/macrozoospermia

Additional NIH Resources

- Eunice Kennedy Shriver National Institute of Child Health and Human Development: How Common is Male Infertility and What are its Causes?
  https://www.nichd.nih.gov/health/topics/menshealth/conditioninfo/Pages/infertility.aspx

Educational Resources

- Cleveland Clinic: Male Infertility
  http://my.clevelandclinic.org/health/articles/male-infertility
- Disease InfoSearch: Infertility associated with multi-tailed spermatozoa and excessive DNA
  http://www.diseaseinfosearch.org/Infertility+associated+with+multi-tailed+spermatozoa+and+excessive+DNA/8663
- Johns Hopkins Medicine: Male Factor Infertility
  http://www.hopkinsmedicine.org/healthlibrary/conditions/adult/kidney_and_urinary_system_disorders/male_factor_infertility_85,P01484/
- MalaCards: spermatogenic failure 5
  http://www.malacards.org/card/spermatogenic_failure_5
- Merck Manual Professional Version: Sperm Disorders
  http://www.merckmanuals.com/professional/gynecology-and-obstetrics/infertility/sperm-disorders
- University of California, San Francisco Medical Center: Infertility in Men
  https://www.ucsfhealth.org/conditions/infertility_in_men/

Patient Support and Advocacy Resources

- RESOLVE: The National Infertility Association: Male Factor Infertility
  http://www.resolve.org/about-infertility/medical-conditions/male-factor.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28macrozoospermia%29+OR+%28%28AURKC%5BTIAB%5D%29+AND+%28sperm%5BALL%5D%29%29+AND+english%5Bla%5D

OMIM

- SPERMATOGENIC FAILURE 5
  http://omim.org/entry/243060

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